Case Report
JCAM

A case of Fahr's disease presenting with seizures

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Abstract

Fahr's disease is a rare idiopathic bilateral and symmetrical calcification of the basal ganglia, thalami, subcortical hemispheric white matter and deep cerebellar nuclei. We report an unusual case of Fahr's disease in a 53-year-old man who presented with generalized seizure in our emergency department. Based on clinical, radiological and endocrinological appearance, the patient was diagnosed with Fahr's disease associated with hypoparathyroidism. Parenteral calcium and calcitriol supplementation were given in the emergency department. The clinical outcome was favorable after the treatment. Our case illustrates that Fahr's disease, though rarely seen, has to be considered in a patient with convulsive state associated with calcifications of the basal ganglia.

Keywords

Fahr's Disease; Hypoparathyroidism; Seizure

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Introduction

Fahr's disease (FD) is a rare entity characterized by symmetrical and bilateral calcifications over the basal ganglia, thalami, cerebellar dentate nucleus or white matter of the cerebral hemispheres [1]. The clinical manifestations of FD vary. General clinical features include movement disorders such as parkinsonism, speech disorders, psychiatric disorders, epileptic seizure, dementia, cerebellar or extrapyramidal dysfunction. Some cases with FD may present without neurological abnormalities [2-3]. It may be sporadic or familial as well as secondary to anoxia, irradiation, systemic disorders, toxins, and disorders of calcium metabolism [4-5]. We report a very rare case of FD due to idiopathic hypoparathyroidism in a 53-year-old man diagnosed by clinical and radiological evidence.

Case report

A 53-year-old male presented to our emergency department (ED) with sudden onset of a generalised tonic-clonic seizure. The oropharyngeal airway was inserted to maintain the unconscious patient's airway. Diazepam was given as the first-line antiepileptic drug, and the seizure was responsive to the treatment. After the patient recuperated consciousness, a neurological examination was performed. Abnormal neurological signs including a mild degree of dysarthria, paresthesia and generalized neuromuscular irritability such as muscle cramps and tetany were found. Latent tetanic convulsions could also be displayed through the elicitation of Chvostek's sign and Trousseau's sign. The patient's past medical history included convulsive episodes which were diagnosed as epilepsy. Laboratory studies including serum calcium 5.2 mg/dl (normal 8.8-10.6 mg/dl), phosphate 7.4 mg/dl (normal 2.5-4.5 mg/dl) and parathormone (PTH) level 3.2 pg/ml (normal 15-65 pg/ml) demonstrated idiopathic hypoparathyroidism. Additional diagnostic laboratory tests including thyroid hormones and vitamins were within normal range. His cranial computed tomography (CT) showed extensive symmetrical calcification of basal ganglia and cerebellum (Figure 1). In addition, bilateral and symmetrical calcifications were also detected over the lateral periventricular areas (Figure 2). A diagnosis of FD associated with idiopathic hypoparathyroidism was made on the basis of the clinical, laboratory and radiological findings. The patient was consulted by a neurologist for a probable seizure, and the patient was treated with an intravenous calcium infusion followed by per oral supplemental calcium and calcitriol, then referred to the endocrinology and neurology departments.

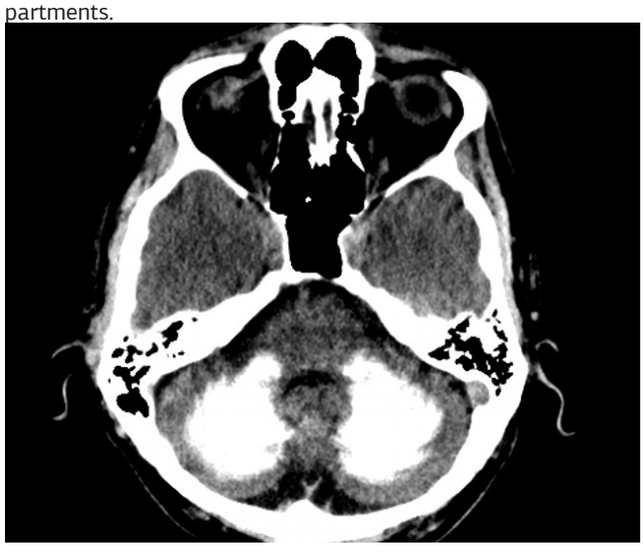

Figure 1. Symmetrical calcification of basal ganglia and cerebellum

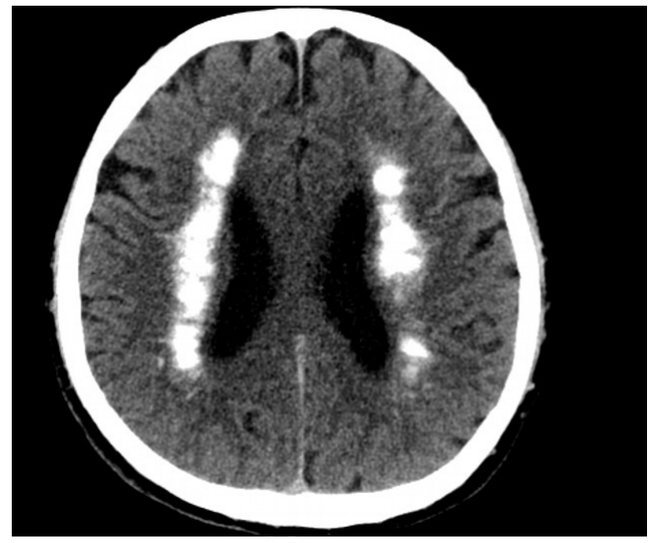

Figure 2. Symmetrical calcifications were also detected over the lateral periventricular areas

Discussion

FD is a rare inherited or sporadic neurological disorder characterized by the presence of bilateral intracranial calcifications with a predilection for the basal ganglia and dentate nuclei. It is commonly associated with endocrine disorders, particularly parathyroid and Vitamin D disturbances [6].

Clinical diagnosis is facilitated by the presence of bilateral and symmetrical calcifications in the basal ganglia in cranial CT. Detection of intracranial calcifications in CT scan is more sensitive compared to skull X-ray or magnetic resonance imaging. It was reported in the literature that in FD, there were symmetrical and extensive calcifications in the white matter of the cerebral or cerebellar hemispheres and basal ganglia [7-8]. As in the present case, the cranial CT findings were typical and consistent with those of FD.

Clinical presentations may be fairly variable. Neurological and psychiatric symptoms, if present at all, are highly variable and include progressive mental deterioration, convulsive seizures, parkinsonism, difficulty in speaking, ataxia, psychosis or affective disorders [1,9]. In the present case, there were no psychiatric symptoms. There were neurological signs such as dysarthria or those of hypoparathyroidism such as latent tetany and seizure.

Hypoparathyroidism has been reported as a cause of FD. It constitutes a very rare group of heterogeneous disorders characterized by hypocalcemia, hyperphosphatemia, normal or increased parathormone secretion, and target tissue resistance to the actions of PTH. Most reported cases of hypoparathyroidism are preceded by or simultaneously present with autoimmune disorders, such as Sjogren's syndrome, Hashimoto's thyroiditis, or Graves' disease [10]. The pathophysiology of this condition is not well-defined, so there is no standard course of treatment. Treatment addresses symptoms on an individual basis [4-5]. Some reports of FD, correction of calcium and phosphate levels may lead to clinical improvement, in particular, the disappearance of epileptic seizures and abnormal movements. As in the present case, after treatment with parenteral calcium and calcitriol supplementation, there were clinical and laboratory improvements.

Conclusion

FD clinical manifestations can vary. It may begin at different ages and have a variety of presentations. The present case is

important because it would appear that there is a few case in the literature of FD presenting with a generalized tonic-clonic seizure. The seizures in such patients may be due to calcium metabolism abnormities and/or dysfunction of cortico-basal connections.

Scientific Responsibility Statement

The authors declare that they are responsible for the article's scientific content including study design, data collection, analysis and interpretation, writing, some of the main line, or all of the preparation and scientific review of the contents and approval of the final version of the article.

Animal and human rights statement

All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. No animal or human studies were carried out by the authors for this article.

Conflict of interest

None of the authors received any type of financial support that could be considered potential conflict of interest regarding the manuscript or its submission.

References

1. Schmidt U, Mursch K, Halatsch M. Symmetrical intracerebral and intracerebellar calcification ("Fahr's disease"). Functional Neurology. 2005;20(1):15.
2. Manyam BV, Walters AS, Narla KR. Bilateral striopallidodentate calcinosis: clinical characteristics of patients seen in a registry. Movement disorders. 2001;16(2):258-64.
3. Lam JS, Fong SY, Yiu G, Wing Y. Fahr's disease: a differential diagnosis of frontal lobe syndrome. Hong Kong Medical Journal. 2007;13(1):75.
4. Verulashvili I, Glonti L, Miminoshvili D, Maniia M, Mdivani K. Basal ganglia calcification: clinical manifestations and diagnostic evaluation. Georgian medical news. 2006; 140:39-43.
5. Baptista MV, Vale J, Leitão O. Striato-pallido-dentate calcifications. Acta medica portuguesa. 1997;10(8-9):563-7.
6. Oueslati I, Khiari K, Bchir N, Abdallah NB. Hypocalcemia and Fahr syndrome in a patient with Graves' disease: Difficult etiological diagnosis. Indian journal of endocrinology and metabolism. 2016;20(6):885.
7. Benke T, Karner E, Seppi K, Delazer M, Marksteiner J, Donnemiller E. Subacute dementia, and imaging correlates in a case of Fahr's disease. Journal of Neurology, Neurosurgery & Psychiatry. 2004;75(8):1163-5.
8. Ogi S, Fukumitsu N, Tsuchida D, Uchiyama M, Mori Y, Matsui K. Imaging of bilateral striopallidodentate calcinosis. Clinical nuclear medicine. 2002;27(10):721-4.
9. Gülsün M, Baykız AF, Kabataş S, Belli H. Fahr syndrome-Three cases presenting with psychiatric signs. 2006; 3(1): 35-40.
10. Murakami T, Nambu T, Morimoto Y, Matsuda Y, Matsuo K, Yonemitsu S, et al. Pseudohypoparathyroidism type II in a woman with a history of thyroid surgery. Internal Medicine. 2014;53(7):743-7.

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